Cystic Fibrosis (CFTR), 165 Pathogenic Variants (2013661)

Tests for 165 pathogenic variants.
Order for the following indications:
- Carrier testing in a healthy patient
- Patient has symptoms of CF
- Patient has family history of CF

Cystic Fibrosis (CFTR), 165 Pathogenic Variants, Fetal (2013662)

0050608—Maternal sample is recommended to rule out maternal cell contamination.

Tests amniocytes for 165 pathogenic variants.
Order for fetal samples with the following indications:
- Both parents have CFTR variants which are known to be included on the panel
- Echogenic or cystic dilatation of the fetal bowel

Cystic Fibrosis (CFTR) Sequencing (0051110)

Detects 97 percent of the more than 2,000 described CFTR gene variants.
Order for patients with symptoms of classic CF or a CFTR-related disorder but without two variants identified by the CF 165 pathogenic variants.

Cystic Fibrosis (CFTR), 165 Variants with Reflex to Sequencing (2013663)

Tests for 165 pathogenic variants. If two CFTR variants are not identified, sequencing is performed to detect 97 percent of the more than 2,000 described CFTR variants.
Order for patients with the following indications if not previously tested using the CF 165 pathogenic variants test:
- Patients with symptoms of classic CF
- Patients with a positive sweat chloride
- Patients with symptoms of a CFTR-related disorder

Cystic Fibrosis (CFTR) Sequencing with Reflex to Deletion/Duplication (0051640)

Tests for more than 2,000 CFTR gene variants. If two pathogenic CFTR variants are not identified by sequencing, deletion/duplication testing is performed; clinical sensitivity is 99 percent.
Order for patients with symptoms of classic CF or a CFTR-related disorder but without two variants identified by the CF 165 pathogenic variants test.
Cystic Fibrosis (CFTR) Deletion/Duplication (0051642)

Tests for large deletions or duplications in the CFTR gene, accounting for 1–2 percent of variants.

Order for the following patients:
- Symptoms of classic CF or a CFTR-related disorder without two identified pathogenic variants following CFTR sequencing.
- To determine carrier status in individuals who have a family member with a large CFTR gene deletion.

Cystic Fibrosis (CFTR), 165 Variants with Reflex to Sequencing and Reflex to Deletion/Duplication (2013664)

Tests for 165 pathogenic CFTR pathogenic variants. If two pathogenic CFTR variants are not identified, sequencing is performed. If two CFTR variants are not identified by sequencing, deletion/duplication testing is performed; clinical sensitivity is 99 percent.

Order for patients with a clinical diagnosis of CF as a cost-effective method for determining the causative variants present.